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00:00.000 --> 00:15.800 Support for Yale Cancer Answers comes from AstraZeneca, a global biopharmaceutical company that is committed to bringing to immuno-oncology to people living with earlier stages of cancer. Learn more at astrazeneca-us.com.

00:15.800 --> 00:46.500 Welcome to Yale Cancer Answers with doctors Anees Chagpar and Steven Gore. Yale Cancer Answers features the latest information on cancer care by welcoming oncologists and specialists who are on the forefront of the battle to fight cancer. This week, it is a conversation about cancer genetics and high-risk patients with Amanda Ganzak, a lead genetic counselor at Smilow Cancer Hospital. Dr. Gore is a Professor of Internal Medicine and Hematology at Yale School of Medicine and Director of Hematologic Malignancies at the Yale Cancer Center.

00:46.500 --> 01:19.700 <vGore>I have to say I hear about genetics with almost every new patient of mine. My patients mostly have leukemia or other related blood malignancies. They always want to know is this something I am going to pass on to my kids. That question is where everybody seems to go. Of course, you see a slice of people who want to know that right, but what do you think about that?

01:19.700 --> 02:01.000 <vGanzak>Yeah. I think it is a very common question that people have, just like anything in a family history whether it is heart disease or diabetes, they want to know am I at risk for this and is my family at risk for this. And the important thing to remember for those with a cancer diagnosis, is only about 5-10% of cancers are hereditary or genetic or that we have genetic testing to be able to understand the risk for them and their family member. So, the vast majority of patients who are diagnosed with cancer will not fall into this category and their relatives will not have a high risk for cancer, but if you fall in that 5-10%, those are the people that we would like to be seeing and testing to help alert their relatives of an increased risk.

02:01.000 --> 02:04.500 <vGore> How would somebody know that they were in that category?

02:04.500 --> 02:26.100 <vGanzak>It is difficult, and I think what we are learning is that there are more people in that category than we once understood before, which is making it more complicated and complex. We used to rely so much on the family history if there were many people with the same types of cancers or related types of cancers, like breast or ovarian cancer in their family, those were patterns that we would look for.

02:26.100 --> 02:36.400 <vGore>So, people know that aunt Tilly had breast cancer and I heard great aunt so and so, and mom had cancer at an early age, like that?

02:36.400 --> 03:20.500 <vGanzak>Those would all be factors that would raise red flags to the majority of physicians who are seeing these patients or the

patient themselves might be raising questions. I think what has become more difficult is that we are learning that not all the family histories look that drastic and there can be more subtle family histories, as an example, let us say the family history is on the dad's side of the family, but there is not a lot of women who would have been at risk for female-related cancer. So, you might not have a strong "family history" like we would be looking for, but it does not exclude the fact that it could still be hereditary. So, those are some of the things that we would evaluate for during a genetic counseling session but might not be as obvious to the clinicians who are seeing these patients in the clinic regularly.

03:20.500 --> 03:49.900 <vGore>Right. And a lot of the cancer susceptibility genes or propensity genes, I am trying to think what the lay word for that would be. The genes that make you at risk for cancer. I hate when we use words like that, it is like oh! God I am talking about the 5 syllable words, they never let my guests get away with that. So, these genes that put you at risk for cancer, not all of them predict for a particular kind of cancer right? I mean, am I right that it may not be just looking for a pattern of breast cancer, could it be looking for a lot of cancers?

03:49.900 --> 04:11.600 <vGanzak>Yeah. It can be dependent on the gene for sure and certain genes have much higher risk than others, but you can see genes associated with breast cancer, colon cancer, ovarian or uterine cancer, pancreatic cancer. We are learning more about the hematologic malignancies and genes related to those now. So really any cancers under the sun, you can sort of find a gene that is linked to it in some way.

04:11.600 --> 04:21.100 <vGore>But a particular gene can also be linked to several right? If you are just looking for a family of just one cancer.

04:21.100 --> 04:29.400 <vGanzak> Correct. Yeah, you are typically seeing 2 or 3 different types of cancers that are clustering together.

04:29.400 --> 05:11.000 <vGore>Right, and I know from my patients again that it is not what they are often looking for, they are looking for blood cancers in my case. You know, it is interesting what you say about the blood cancers, one of my colleagues in Chicago whom you may be familiar with, Dr. Lucy Godley, really made us a thing out of discovering familial leukemia-associated genes and she came and gave us a talk recently and it was something like if you have leukemia and you have 1 or 2 first-degree relatives with any cancer, then the chance of finding a gene was something like 20 or 25%. I mean, really surprisingly high.

05:11.000 --> 05:32.200 <vGanzak>Yeah. I do not know that literature quite as well, I will be honest, but when we are starting to see multiple people with leukemias in a family, that starts to reach that pattern concerning of something hereditary. Now whether or not we can find an identifiable gene that is linking all those together, that is different, but there certainly have been many genes in the last 5-10 years that we really have found in some of these hematologic families, particularly with the leukemias.

05:32.200 --> 05:36.900 <vGore>So, how do people come to you. Do people self-refer or their oncologist refers?

05:36.900 --> 06:02.500 <vGanzak>The vast majority of patients come to us from one of their physicians, whether that be a primary care, gynecologist, gastroenterologist or from the cancer center - so from their oncologist or their surgeon might be referring them, most of them end up that way, referred to our center. We do have some self-referrals especially if family members have been seen and they are coming in for testing, they might just be cold calling us for an appointment, but the majority come from some healthcare provider that they are seeing.

06:02.500 --> 06:07.400 <vGore>And do most of the people who are coming to see you have a diagnosis of cancer or some of them are worried about cancer?

06:07.400 --> 06:18.200 <vGanzak>I would say it is a good split. They are very worried coming in based on their family history of cancer and we are seeing a large majority of people with cancer.

06:18.200 --> 06:23.100 <vGore>I mean, I came into the clinic that way as a matter of fact.

06:23.100 --> 06:35.000 <vGanzak>And I think it is a good thing. I would rather see someone and either be reassuring or say to them, you know this is great that you came in, I am glad we can take this opportunity to review the history and get you the care that is most appropriate.

06:35.000 --> 07:04.900 <vGore>So, how does that work? I come in because in my case, I was concerned about whether there was a familial risk of pancreas cancer due to my mother and her sister, which in my understanding 2 relatives in that same generation was enough to be concerned. And in my case, I was seen by a genetic counselor I think primarily, is that the usual pattern?

07:04.900 --> 08:19.500 <vGanzak>Yeah. The usual pattern is a patient will come in, they will meet with a genetic counselor, we typically meet with patients for about 1 hour. We ask pretty detailed questions about the family history. So, we let patients know in advance, ask some of your relatives questions before you come in, things like how old was grandma when she had breast cancer, how old was uncle Mike when he had pancreatic cancer. So, that when we meet, we have as much detailed information as possible to make our assessment. I am looking at the number of people in the family with cancer, the types of cancers, the ages when they were diagnosed, but also might be looking at who does not have cancer. You know, how does that ratio play out within the family. If I am starting to see more people with cancer than without, then that is going to be something more striking for an inherited risk factor. We will go through what are genes, how do they cause cancer, what does genetic testing look like, how might we use these results to guide cancer screening and decision making in that regard. And so, within that hour, we are helping patients navigate and make an informed choice about whether genetic testing is indicated, whether

they want to move forward with it and what exact tests do they want to choose moving forward with the options they have.

08:19.500 --> 08:25.800 <vGore> So, are there genetic tests for every situation?

08:25.800 --> 09:31.500 <vGanzak>No. There are not. And that can be difficult I think when there are conversations in a family that say, this looks pretty hereditary, but I do not know what gene it is yet, come back or call us in 5 or 10 years, that is not always easy to have that conversation with a patient, but it happens. We made advances and we have learned so much and even in the past over 10 years that I have been doing this, the number of genes we look at has exponentially changed and so it will continue to do so. I think one of the very important things I tell patients when they leave our office or give them results of genetic testing is, this is not over just because we did this test and we have gotten our results, you might want to continue to keep in contact with us - family history has changed, there are new diagnosis that are made within a family that could change the assessment and the genes we might want to look at, we learn more about different genes that we might want to test for in the future. So, well, it feels like a snap shot in time when they meet with us. We want to continue to evaluate that over time to make sure that if anything has changed that we have accounted for that and our recommendations are changed accordingly.

09:31.500 --> 09:35.600 <vGore>How do you do the testing. Do you actually have to do biopsies of various tissues?

09:35.600 --> 10:02.100 <vGanzak>No we do not. That is probably one of the biggest questions we get walking in. There are 2 things people ask me - is this going to hurt and how much is this going to cost me? The first one is. it is not going to hurt. Usually, we do it through a blood sample. It is a pretty small amount, like a tablespoon size of blood that we would take and the testing can also be done through a saliva sample, so if someone really does not want to have their blood drawn, then we can use saliva sample. They just spit into a tube for us.

10:02.100 --> 10:04.800 <vGore>So, that's easy. Now, lets talk about the payment part.

10:04.800 --> 10:45.700 <vGanzak>That is definitely very complex. And as a moving target, the testing companies out there, there are many different companies that offer genetic testing way more than there used to be and the cost has drastically come down. So, it really makes genetic testing a lot more affordable and doable for people where they might not have had that opportunity before. So, our conversations are even if you do not meet your insurance criteria, you could have the option to pay out of pocket, costs are around 250 dollars right now if patients decide to self-pay. Now, that is not nominal, that is still a lot of money for a lot of people.

10:45.700 --> 10:50.700 <vGore>It is a lot different than 5000 like it used to

be.

10:50.700 --> 11:16.100 <vGanzak> Exactly. So, the insurance companies are definitely giving us more pushback, we have to prove a lot more - the number of people in the family who have cancer, why is this indicated, how is it going to change medical care and those justifications take a lot more time than they used to. So, we are kind of caught in the middle as genetic counselors to justify why this test is needed, how we will use that information to hopefully get that test paid for.

11:16.100 --> 11:36.800 <vGore>What is the consequence, lets say you are screening somebody and you find a gene that puts somebody at risk for some kind of cancer or several kinds of cancers, now what?

11:36.800 --> 12:52.500 <vGanzak>That is probably a bulk of our patients who come through with inherited risks. They are a young individual who now is facing this lifelong, potentially, action plan. We like to think of ourselves as the quarterback of their care through the cancer prevention center. We want to be able to help coordinate the appropriate cancer screenings for that individual moving forward. As an example, if someone had a condition like Lynch syndrome, which increases the risk for colon cancer, other gastrointestinal cancers and for women - gynecologic risk, we have a lot of people to coordinate with - we have to make sure they are connected with the gastroenterologist who will screen their colon and their upper GI tract more regularly. For women, we have to connect them with their gynecologist and making sure that they are getting appropriate screening, and it can be overwhleming for people, this is a lot of appointments, it is a lot of testing that they have to do, it can be very overwhelming in the beginning to coordinate all of that. And so, we really hope that our program is there to help some of that, to take some of that burden off the patient, make it a little bit easier, set them up with those appointments and get them in a good routine so they know what to do every year moving forward. But it is a lifelong commitment that these individuals are having to make due to these risks.

12:52.500 --> 12:57.800 <vGore>So, that is what I was going to ask, it is not like you get screened once or twice and you are done right?

12:57.800 --> 13:07.700 <vGanzak>No, if we use Lynch syndrome as our example again, these individuals are getting colonoscopies every single year from when they are 20 to 25 years old onwards.

13:07.700 --> 13:11.600 <vGore>That is a lot of colonoscopies.

13:11.600 --> 13:15.900 <vGanzak>A lot of colonoscopies for a 20-year-old, 25-year-old, try to convince them to come in every year, it is hard.

13:15.900 --> 13:32.900 Medical Minute Support for Yale Cancer Answers comes from AstraZeneca, providing important treatment options for patients with different types of lung, bladder, ovarian, breast, and blood cancer. More information at astrazeneca-us.com.

13:32.900 --> 14:15.900 This is a medical minute about melanoma. While melanoma accounts for only about 4% of skin cancer cases, it causes the most skin cancer deaths. When detected early, however, melanoma is easily treated and highly curable. Clinical trials are currently underway to test innovative new treatments for melanoma. The goal of the specialized programs of research excellence in skin cancer or SPORE grant is to better understand the biology of skin cancer with a focus on discovering targets that will lead to improved diagnosis and treatment. More information is available at YaleCancerCenter.org. You are listening to Connecticut Public Radio.

14:15.900 --> 14:57.000 <vGore>Welcome back to Yale Cancer Answers. This is Dr. Steven Gore. I am joined tonight by my guest Amanda Ganzak. We have been discussing cancer genetics and high risk patients. So, Amanda, before the break, we were talking about getting your insurance pay for your screening and the other question that I had about insurance is the one that for a long time people had worried a lot about, so you get diagnosed with a gene that is associated with something like the Lynch syndrome gene or the BRCA genes that many women know about for breast cancer risks and others, and people worry about having higher premiums or being denied insurance. What is the story with that nowadays?

14:57.000 --> 15:46.600 <vGanzak>I still think it is a very big concern that patients have, how will this genetic information be used against me? And there are laws both federally and state wide that have been put in place to protect individuals. The federal law is called the genetic information nondiscrimination act, or GINA for short. This law has protections for those individuals who are unaffected, who have been identified with genetic risk and that does not just mean cancer, it can be any genetic disease, but unfortunately, if someone has a cancer diagnosis, they are now affected and GINA no longer protects against that, so insurance could use potentially the results of genetic testing to make decisions, but they are also using their cancer diagnosis to make decisions about premiums, which is probably more weighted than their genetic test results.

15:46.600 --> 15:53.700 <vGore> The affordable care act prevents from them from being denied on either of those right?

15:53.700 --> 16:07.500 <vGanzak>Correct. I think it can get a little nuanced, this is certainly something we talk about with patients, but it is not a blanket statement everyone is protected and cleared and genetic testing could be used, but it is likely not used to make decisions at this point.

16:07.500 --> 16:20.200 <vGore> And I also assume that if somebody is going to need to have annual colonoscopies or mammographies or whatever it may be, they are going to need to explain to their insurance company why they need so many colonoscopies?

16:20.200 --> 16:39.700 <vGanzak>Exactly. It needs to be somewhere quoted that they have an increased risk for some reason to get the colonoscopy every year. So, that genetic diagnosis is our way to get that covered but many patients

are worried that having that in their record will flag that to their insurance. So, it is a double-edged sword I think that we get into.

16:39.700 --> 16:56.800 <vGore>I mean, I think societally it is really interesting because these people always existed and so, in someways, we can potentially save the insurance companies and society money by doing appropriate screening and not dealing with advanced cancer I would think, as well as saving lives, that seems like a good thing.

16:56.800 --> 17:31.100 <vGanzak>Yeah, and that is what I try to tell my patients, when you move forward with risk-producing surgery, you have your ovaries removed and you essentially eliminate that risk, you are at less risk than the general population woman is, and you would think that insurance would see the benefit of that. And I think many of them do, they are covering these risk-producing surgeries, they are covering these increased surveillance, things that are considered standard of care with the genetic diagnosis, so it is getting recognized from the insurance company, these are important things that we need to do and in the end, yes it probably saves money by delaying or totally avoiding a cancer diagnosis in the first place or at least an advanced one.

17:31.100--> 17:48.900 <vGore>Do you think that the coming out of certain public figures, I think it was Angelina Jolie with her BRCA genes and the dramatic prophylactic surgery she had, has that increased the conversation or nobody remembers that anymore?

17:48.900 --> 18:34.000 <vGanzak>In our world of genetics, we like to call that the Angelina Jolie effect. We definitely saw an increase of referrals and increase of calls and increase of interest from individuals coming in asking, you know my family looks or sounds a lot like Angelina Jolie's is this something that I should be worried about? When I talk about the BRCA genes, if I mention Angelina Jolie, it makes sense and so it helps. I think it brings light to something that a lot of people were not comfortable thinking about or talking about with their family and it brings it to the masses and I think it has benefits. So, while it made us really busy in the clinic, it is in the end I think a really good thing that people are starting to question their family history more than they might have otherwise.

18:34.000 --> 19:04.700 <vGore>It is interesting, I know that when Katie Couric had the onscreen colonoscopy that was very good for raising colon cancer awareness and Robin Roberts who had a stem cell transplant from myelodysplastic syndrome, I think I do not wish illness on any celebrities, but I really do respect those celebrities who use their experience as an opportunity to give back given what they have gotten from the public to the public, that is pretty cool.

19:04.700 --> 19:04.100 <vGanzak>I completely agree.

19:04.100 --> 19:14.600 <vGore>What percent of people who come to see you actually turn out to have something, have a bad gene?

19:14.600 --> 19:39.900 <vGanzak>I think we are a little biased because the people who come to us probably have a pretty strong family history, but I would say about a quarter of our patients who move forward with genetic testing might end up to have something hereditary in some way, whether that is identifiable or not, but once again if we look across the population, any cancer, about 5-10% of it will be hereditary. So, we are sort of narrowing in on those individuals who probably have more risk than others.

19:39.900 --> 19:55.400 <vGore>Kind of puts you in an emotionally tenuous position where it is more interesting for your job that somebody turns out to have something, but you do not really want anybody to have anything.

19:55.400 --> 20:40.600 <vGanzak>You know, the conversations have definitely become more interesting as there have been more genes discovered and things are changing. So, I am having these families where we did not have genetic testing years ago and now, we have that gene and it totally flips the family upside down, thinking that they did not have anything and now we are able to answer those questions. So, it is definitely very interesting, but yeah you do not wish it upon anyone when they walk through the doors, but at the same time when you do identify it, it gives an answer and I think it is an answer that some families have been searching for, for a long time - why did this happen to us and it can always be a sense of relief, it is nothing we did, it is nothing we ate, no bad decisions coming back to us, it is just something we were born with and we have to deal with, but now we know and we can.

20:40.600 --> 21:31.000 <vGore>I remember a patient that I had, I think it is back from my Johns Hopkins days, which is at least 6 years ago, and there was a family with a very impressively peculiar history of many patients with the some weird leukemia, it had to be something and I sent it to my colleague in Chicago that I mentioned and they were able to identify it and now I happen to be mentioned on a paper that grew out of that, but I think it was very empowering to the patient to know that there was somebody looking for what the family knew was a problem, so what I was going to ask you about was, are you working with any research groups, do you refer people to research groups if the story is so that it has got to be something and you do not find something, what do you do or where do you refer people?

21:31.000 --> 22:24.400 <vGanzak>Wonderful. We actually have our own research repository within Yale. So, we offer all of our patients the opportunity to enroll to our research repository and what we would do is collect a blood sample or a DNA sample through saliva, have them consent obviously to use that sample and over time, that gives researchers the opportunity to say of all our patients with leukemia or all our patients with colon cancer under 50, let us take those samples and go beyond the genes that we have already looked at and know about what else could it be. So, we use samples in that way. We also can send samples, our coordinate with other institutions if there is a research group somewhere else that is looking at these particular types of families, ask them if they be interested in enrolling as well. So, we sort help navigate both

of that, both in-house as well as external.

22:24.400 --> 22:31.100 <vGore> And in such cases, do you actually get samples on family members or if there is nothing to measure, you do not do that at that time?

22:31.100 --> 23:15.200 <vGanzak>It sort of depends. If we have a family where there is a lot of affected individuals and we have not found the inherited risk, collecting samples from those affected individuals allow research just to have more power in numbers to make the linkage within the family, so a family like that would be someone that we really would try to collect as many samples as we could. If we have not identified anything and there is not really strong family history that is probably not the one that we would extend offering samples to be collected. So, it is sort of a case by case basis and we present our families to our medical directors weekly and anytime that there might be a good research family, the genetic counselor might go back and coordinate that with the family.

23:15.200 --> 23:37.600 <vGore>Got it. I just kind of interested in your story. You say you have been in this about 10 years, did you go into genetic counseling with a particular interesting cancer or it is just genetics or how does one come to this career, it is not something, maybe they talked to you about in freshman biology class I do not know, but it is not on your list like you want to be a teacher, doctor, fire person, you know.

23:37.600 --> 24:49.900 <vGanzak>Yeah definitely not. I mean, most people do not even know what it is when you mention it. I in particular was really interested genetics. I actually took a genetics class in high school, so for me I sort of always liked biology and genetics and it just so happened that mom has a BRCA1 mutation. So, for me it is personal. I saw her diagnosed with breast cancer at a young age, I saw her go through the process of meeting with a genetic counselor and learning of this risk and the value that I saw that the genetic counselor brought to her was something that really highlighted to me - this is a very interesting field and something I think could work well for me. It is a really great combination between really having to keep up-to-date all the time with genetics and science and it is ever changing, which is very exciting and you know no two days are the same, but it also allows me to meet with patients one on one for a long period of time; I mean, who has an hour that they get to spend in the clinic with patients, it is a very rare field that you get to spend that much time and connect with patients and so for me, it kind of had the dual benefit of both and I can take my own personal experience and put that into my work and I think it brings it to a different level than maybe many other genetic counselors might have the opportunity to do.

24:49.900--> 24:57.600 <vGore>And so, you were really always going into cancer. Wow. And I assume you have to do post baccalaureate training.

24:57.600 --> 25:31.500 <vGanzak>Yeah. It is a 2 years master's program. The field has almost doubled in size since I have started. So, it is definitely a field that is growing and expanding, the need for our services is greater than the number

of genetic counselors we have. There are various specialities from prenatal, pediatric, cancer, but it is expanding into neurology, cardiology, even psychology, you know looking at some of these more pscyh disorders and could there be a connection there. So, it really is expanding across the field of medicine.

25:31.500 --> 25:40.500 <vGore>And when you do your master's training, is there a way to subspecialize then and enhance your knowledge about cancer or pscyh or whatever it may be?

25:40.500 --> 26:14.300 <vGanzak>Yeah, so the programs are designed in such a way that we have to rotate through the core specialities. So, prenatal, pediatric and cancer are considered core. So, we have to rotate through those clinics, but there are opportunities that if you really like cancer, you might choose a second rotation in cancer again to get that added speciality. So, you can sort of specialize, I guess I would say if you could arrange it that way, but you really are getting exposed to everything. We take a board exam when we graduate and it covers everything, so we have to make sure we have that good foundation.

26:14.300 --> 26:42.900 <vGore>Wow. That's a lot to learn. I certainly cannot even keep up with it in my field, so in terms of keeping up with it, I know that the scientific laboratory-based platforms by which these things are tested change all the time and particularly when it is not one certain gene you are looking for, and so do people have to get retested if you do not find anything or can you keep their sample and re-run it, how does that work?

26:42.900 --> 27:32.800 <vGanzak>Yeah. It can depend on where they had their initial testing. For most commercial companies, if they had testing greater than just a couple of months ago, that a new sample is required to have more genes tested. Here at Yale, we have the opportunity to go through the Yale Diagnostic Laboratory and they will keep a sample, so we might have the opportunity if it has been within a couple of years to re-run that sample. So, it can depend. We do offer more testing to patients if it has been long enough, if there have been new genes and they were not tested before, they would be given the opportunity to come back in, review those genes and what benefit it could have to them and offer testing if necessary. So, we certainly are seeing that now. We offer women and men who have had prior BRCA testing only and no other breast cancer genes tested, saying why do not you come back in, there is more that we could do now.

27:32.800--> 27:40.400 <vGore>So, do you actually call people back or do you set them up for a followup, so how does that work?

27:40.400 --> 28:03.400 <vGanzak>We leave a little bit of the onus on the patient to contact us. We have so many patients come through that it would be a never-ending list of people that we would be calling, we probably spend more time doing that than actually seeing them. So, we tell patients all of the time if has been a couple years and you are still worried or have questions, call us. We review the results that previously had been taken in place, we look at that and say more testing is warranted, why do not you come back in.

28:03.400 --> 28:15.500 <vGore>Gotcha. And so, just in the last couple of minutes we have, I suspect that a lot of people are using some of the geneology DNA things to do kind of a cheap screen, what do you think about that?

28:15.500 --> 29:00.800 <vGanzak>I mean we caution patients using what we would call recreational genetics. So, there are limitations around these tests. They have false positives, they have false negatives, they can have psychological implications, things like learning what you think is risk that turns out not to be real risk or that there is an opportunity. There can be limitations to these testing and real impact, so from our National Study of Genetic Counselors, our position statement has sort of been do it for fun but with caution, and know its risks and its limitations, use a genetic provider who can interpret these results and help you along the way, but it might not be the test that we would use clinically to guide medical management.

29:00.800 --> 29:24.100 Amanda Ganzak is the lead genetic counselor at Smilow Cancer Hospital. If you have questions, the address is canceranswers@yale.edu and past editions of the program are available in audio and written form at YaleCancerCenter.org. We hope you will join us next week to learn more about the fight against cancer here on Connecticut Public Radio.